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In the paper by Mousavi, A. S. et al., the authors explore the development of a PCR-based method for identifying fetal sex during in vitro fertilization (IVF) cycles as a potential alternative to traditional invasive procedures used for preimplantation genetic diagnosis (PGD). Preimplantation genetic diagnosis is commonly used to detect X-linked diseases, where sex determination of embryos becomes critical for couples at risk of passing on such disorders. This study aimed to discriminate between male (XY) and female (XX) embryos by detecting specific sex chromosome sequences in the culture medium, utilizing a quantitative polymerase chain reaction (qPCR) approach.

The authors collected spent culture medium samples from 120 embryos at two stages, Day 3 and Day 5, and extracted cell-free DNA using a modified phenol-chloroform solution. The sex determination was performed by targeting the SRY, TSPY, and AMELOGENIN genes using qPCR. The results from qPCR were compared with those obtained from traditional PGD methods based on array comparative genomic hybridization (CGH). The sensitivity, specificity, positive predictive value, and negative predictive value for determining fetal sex using the qPCR method were 100%, indicating a high accuracy in identifying sex without the need for embryo biopsy. The findings suggest that this non-invasive method could potentially replace invasive procedures, offering a safer and more cost-effective approach to PGD, particularly for sex-linked diseases.

The study further highlights that DNA release into the culture medium increased with embryo development stage, making Day 5 samples more reliable than Day 3 samples for sex determination. Although the study achieved promising results, the authors emphasize the need for further research involving larger sample sizes to validate the method's clinical reliability before widespread adoption.

Work Cited

Mousavi, A. S. et al. Development of a PCR-based method to identify fetal sex during IVF cycles. *Zygote*, https://doi.org/10.1017/S096719942400011X (2024).